Friday 12 April 2024

EUROPEAN REFERENCE NETWORKS
FOR RARE, LOW-PREVALENCE AND COMPLEX DISEASES
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ERN GUARD-Heart

Gateway to Uncommon And Rare Diseases of the Heart

ERN GUARD-HEART BIMONTHLY NEWSLETTER YEAR 2024 NUMBER 2

Meetings for patients

Amsterdam, 09-04-2024

Until 2027, each ERN year there is budget reserved for each (member/ affiliated partner) country to organize a meeting for patients. For this year already 21 meetings in 10 countries are planned, but there are still possibilities for extra meetings. Please don't hesitate to contact the ERN management office for information, possibilities and requests. Below a picture from a meeting in Hungary, in 2023, where a new meeting (Cardiomyopathy Patient Day) will be organized again soon. ♥

Patient meeting in Szeged, Hungary, 2023

ECRD 2024 - Brussels

The European Conference on Rare diseases & Orphan Products (ECRD) is the largest, patient-led, rare disease policy-shaping event held in Europe. By bringing together people with rare diseases and patient advocates, policy makers, healthcare industry representatives, clinicians, regulators and Member State representatives, EURORDIS harnesses the power of this extensive network to shape goal-driven rare disease policies of the future.

With over 1000 participants, the Conference is an unrivalled opportunity to network and exchange invaluable insights within the rare disease community. Through collaborative efforts, these discussions culminate in clear policy recommendations that can influence both EU and national policies.

The next ECRD, a fully hybrid conference, will take place on 15 & 16 May 2024 online and at The Square in Brussels. Visit the following website for the program and registration: https://www.rare-diseases.eu/

EURORDIS
RARE DISEASES EUROPE

PROMS – we need you!

Brussels, 05-04-2024

Request to participate in Our Research on Quality of Life in Inherited Cardiac Conditions! ✳

As presented during the board meeting in Paris, research on the quality of life (QoL) among patients with inherited cardiac conditions (ICC) is limited. Existing evidence from our systematic review indicates that most studies rely on generic measures of health-related QoL, which may not adequately capture ICC-specific factors. The current study aims to develop a disease-specific patient-reported outcome measure to address this gap.
Using a rigorous qualitative methodology, we have embarked on focus group discussions and expert interviews to glean invaluable insights, subsequently informing the construction of our novel instrument. These efforts have not only identified critical QoL domains within the overarching theoretical framework but have also elucidated a distinctive theme, "genetic well-being," encapsulating nuanced constructs germane to ICC patients.

WE NEED YOUR PARTICIPATION!

Saar van Pottelberghe

To select eligible participants aged 18 years and above, diagnosed with ICCs such as Brugada syndrome, Long QT syndrome, and various cardiomyopathies, to partake in the validation of our questionnaire.

Your involvement will be pivotal in advancing our understanding of QoL in ICC patients, potentially leading to the development of more effective interventions and enhancing patient care. Your participation could make a real difference in the lives of ICC patients.

For those interested, don't hesitate to contact us at saartje.vanpottelberghe@uzbrussel.be to receive detailed information and participate in this significant endeavor.

We deeply appreciate your interest and willingness to contribute to this research. Your invaluable contribution is shaping the landscape of ICC patient care. Join us in this meaningful pursuit today! ♥

Meeting for European Patient Advocacy Groups in Paris

Paris, 22 March 2024

At Friday 22 March, the Patient Advocacy Groups (ePAGs) from ERN GUARD-heart organized a physical meeting in Paris, to design new plans and attend the ERN GUARD-Heart board meeting, which was held simultaneously. At the Board meeting, the ePAGs joined some sessions and presented an update of their activities. Nine representatives (from France, Germany, Spain, Ireland, Italy, Finland, Netherlands, and Portugal) were on-site and enjoyed to meet each other, and the ERN board members. ♥

European Reference Networks

ERN-Board Meeting and dinner 21-22 March 2024 in Paris

Paris, 22 March 2024

On Friday 22 March, the ERN Board members, affiliated partners and representatives from the European patient advocacy groups participated in the 15th ERN Board Meeting, which was organized in Paris. With 32 physical attendees and about 20 online, each HCP was able to attend. It was a nice and interactive meeting, with important topics discussed and strategic decisions made. On Thursday evening 21 March, the spring started with a very nice ERN dinner in restaurant 'Le Bleu Train' in Gare de Lyon. Thanks to the group of Philippe Charron for their help with the organisation and thanks to all the attendees for their active participation. ♥

Who are our ePAG's?
Sophie Muir - UK

Amsterdam, 30-03-2023

Following the finding of a CACNA1C variant in my middle son Calvin after nearly a 10-year diagnostic odyssey I registered Timothy Syndrome Alliance (TSA) as a charity. Our mission is to improve the diagnosis, treatment and care of individuals impacted by CACNA1C worldwide and to support them and their caregivers. We are not only raising awareness but growing our community at the same time.

Being a member of the ERN GUARD-HEART Patient Advocacy Board is an amazingly important collaboration addressing the unique challenges faced by those with rare and complex diseases of the heart, which includes CACNA1C. Now if CACNA1C is making you think you've heard of it before it's most likely because you have heard of Timothy Syndrome (TS), a multisystem disorder caused by a specific variant in the CACNA1C gene that presents notably with a prolonged QT interval.

Having grown our community from 43 known living individuals in 2019 when TSA was registered to the 160+ CACNA1C individuals we have connected with today (38 individuals in 2023 alone) we know there to be a wide range of changes outside the pathognomonic p.G406R protein change of Timothy Syndrome. CACNA1C-related disorders are now known to be a large spectrum encompassing neurologic-only, cardiac-only, and mixed neurologic and cardiac symptoms.

CACNA1C variants are associated with an increased risk for abnormal heart rhythm (arrhythmia), often characterized by a prolonged QTc interval on electrocardiogram (ECG).

Other forms of arrhythmia have been reported with CACNA1C variants. A CACNA1C variant can also predispose to structural heart problems, like thickening of the heart wall (left ventricular hypertrophy or hypertrophic cardiomyopathy) or congenital heart defects. Some individuals with a CACNA1C variant can present with neurologic problems and/or physical differences. These may include fused fingers or toes (syndactyly), facial differences, hair and teeth anomalies, predisposition to low blood sugar (hypoglycaemia), developmental delays, intellectual disability, autism spectrum disorder (autistic features), incoordination, ataxia, hypotonia, attention deficit hyperactivity disorder (ADHD), and/or seizure disorder (epilepsy). Our dedicated Scientific Advisory Board helps steer the research efforts of the charity offering expertise on scientific and clinical developments. We are fortunate to have internationally leading researchers and medics serving on the board chaired by Dr. Jack Underwood, Cardiff University including Dr. Anwar Baban, ERN GUARD-Heart (Paediatric Cardiology and Arrhythmia/Syncope Units, Bambino Gesù Children Hospital and Research Institute, IRCCS, Rome).

Together in June, we gathered our global CACNA1C community for our virtual language-accessible conference. By embracing the digital platform, everyone, regardless of their geographic location, had the opportunity to join us, with the presentations from members of our Scientific Advisory Board and guest speakers available post-event along with multi-language transcripts.

The conference served as an opportunity for CACNA1C individuals, families, caregivers, researchers, scientists, healthcare professionals, advocates, and supporters to come together where we collectively shared current knowledge, and ongoing studies, exchanged ideas, and fostered collaborations to help shape the future of CACNA1C research, improved diagnosis, and care.

Collective efforts sharing knowledge such as this conference will continue to improve the lives of those affected by CACNA1C.

Who are our ePAG's? Sophie Muir

Collaboration between healthcare professionals and patients is also strengthened by the essential role of European Reference Networks (ERNs) and the voice of patients in European Patient Advocacy Groups (ePAGs). This partnership allows doctors and patients to exchange life-saving knowledge and expertise without the need for travel. We are delighted to be a part of it.
Due to our conference presentations (available in 14 languages) including work not yet published please email sophie@timothysyndrome.org to request access if you'd like to learn more. Please also engage with our social media channels to amplify our impact. For The Love Of CACNA1C was our February campaign with personal messages of hope, support and signposting aimed at individuals, families and carers who have not found us yet.
www.timothysyndrome.org

Locum Consultant in Paediatric Inherited Cardiology

Great Ormond Street Hospital for Children NHS Trust

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Link to advert for Locum Consultant in Great Ormond Street Hospital for Children NHS Trust

We are keen to receive applications from enthusiastic and expert colleagues!!

Juan Kaski

NHS Great Ormond Street Hospital for Children NHS Foundation Trust

Latest ERN GUARD-Heart Publication (s)

1. Kaski JP, Norrish G, Gimeno Blanes JR, Charron P, Elliott P, Tavazzi L, Tendera M, Laroche C, Maggioni AP, Baban A, Khraiche D, Ziolkowska L, Limongelli G, Ojala T, Gorenflo M, Anastasakis A, Mostafa S, Caforio ALP; EORP Paediatric Cardiomyopathy Registry Investigators. Cardiomyopathies in children and adolescents: aetiology, management, and outcomes in the European Society of Cardiology EURObservational Research Programme Cardiomyopathy and Myocarditis Registry. Eur Heart J. 2024 Mar 1:ehae109. doi: 10.1093/eurheartj/ehae109. Epub ahead of print. PMID: 38427064.
2. Engele LJ, González-Fernández V, Mulder BJM, Ruperti-Repilado FJ, Abia RL, van der Vlist K, Buendía F, Rueda J, Gabriel H, Schrutka L, Bouchardy J, Schwerzmann M, Possner M, Greutmann M, Gallego P, Ladouceur M, Jongbloed MRM, Tobler D, Dos L, Bouma BJ. Decreased clinical performance in TGA-ASO patients after RVOT interventions; a multicenter European collaboration. Int J Cardiol. 2024 Apr 5:132027. doi: 10.1016/j.ijcard.2024.132027. Epub ahead of print. PMID: 38583591.
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6. Butera G, Piccinelli E, Kolesnik A, Averin K, Seaman C, Castaldi B, Cuppini E, Fraisse A, Bautista-Rodriguez C, Hascoet S, D'Amore C, Baruteau AE, Blasco PB, Bianco L, Eicken A, Jones M, Kuo JA, Rajszys GB. Implantation of atrial flow regulator devices in patients with congenital heart disease and children with severe pulmonary hypertension or cardiomyopathy-an international multicenter case series. Front Cardiovasc Med. 2024 Jan 15;10:1332395. doi: 10.3389/fcvm.2023.1332395. PMID: 38288053; PMCID: PMC10822980..

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